California Newborn Screening Program
Conditions Currently Identified

I. Primary (disorders screened for):

- Phenylketonuria (PKU)
- Defects of biopterin co-factor biosynthesis (4)
- Classical galactosemia
- Primary congenital hypothyroidism
- Sickle cell anemia (Hb S/S disease)
- Sickle C disease (Hb S/C disease)
- Sickle D disease (Hb S/D disease)
- Sickle E disease (Hb S/E disease)
- Hb S/ hereditary persistence of fetal hemoglobin (Hb S/HPFH)
- Sickle cell disease variant (Other sickle cell disease, Hb S/V)
- Hb S/ $Beta^0$ thalassemia
- Hb S/$Beta^+$ thalassemia
- Hb H disease
- Hb H/ Constant Spring disease

II. Secondary (disorders in which some cases are also identified):

- Variant hyperphenylalaninemia
- Benign hyperphenylalaninemia
- Duarte galactosemia (D/G)
- Variant hypothyroidism
- Transient hypothyroidism
- Beta thalassemia major
- Hb E/ $Beta^0$ thalassemia
- Hb E/$Beta^+$ thalassemia
- Hb E/ Delta Beta thalassemia
- Hb C/ $Beta^0$ thalassemia
- Hb C/$Beta^+$ thalassemia
- Hb D/ $Beta^0$ thalassemia
- Hb D/$Beta^+$ thalassemia
- Hb Variant/ $Beta^0$ thalassemia
- Hb Variant/$Beta^+$ thalassemia
- Homozygous Hereditary persistence of fetal hemoglobin (HPFH/HPFH)
- Alpha thalassemia major
- Hb H with other variant point mutations
- Hb E disease (Hb EE)
- Hb C disease (Hb CC)
- Hb D disease (Hb DD)
- Other hemoglobinopathies (Hb Variant/Variant)